

SOX10 (https://www.gephebase.org/search-criteria?/and+GeneGephebase=~SOX10~#gephebase-summary-title)	Gephebase Gene	GP00001068	GepheID
Published	Entry Status	Martin	Main curator

	Trait Category		
Morphology (#gephebase-summary-title)			
	Trait		
Coloration (feathers) (#gephebase-summary-title)			
	Trait State in Taxon A		
Columba livia - blue/black			
	Trait State in Taxon B		
Columba livia - recessive red e2 allele			
	Ancestral State		
Taxon A			
	Taxonomic Status		
Domesticated (#gephebase-summary-title)			
Taxon A		Taxon B	
	Latin Name		Latin Name
Columba livia #gephebase-summary-title)		Columba livia #gephebase-summary-title)	
	Common Name		Common Name
rock pigeon		rock pigeon	
	Synonyms		Synonyms
Columba livia domestica; rock pigeon; carrier pigeon; domestic pigeon; rock dove; Columba livia Gmelin, JF, 1789		Columba livia domestica; rock pigeon; carrier pigeon; domestic pigeon; rock dove; Columba livia Gmelin, JF, 1789	
	Rank		Rank
species		species	
	Lineage		Lineage
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Sauropsida; Sauria; Archelosauria; Archosauria; Dinosauria; Saurischia; Theropoda; Coelurosauria; Aves; Neognathae; Columbiformes; Columbidae; Columba		cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Sauropsida; Sauria; Archelosauria; Archosauria; Dinosauria; Saurischia; Theropoda; Coelurosauria; Aves; Neognathae; Columbiformes; Columbidae; Columba	
	Parent		Parent
Columba () - (Rank: genus) https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=8931)		Columba () - (Rank: genus) https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=8931)	
	NCBI Taxonomy ID		NCBI Taxonomy ID
8932 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=8932)		8932 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=8932)	
	is Taxon A an Intraspecies?		is Taxon B an Intraspecies?
No		No	

Sox10	Generic Gene Name	UniProtKB Mus musculus
		Q04888 (http://www.uniprot.org/uniprot/Q04888)
gt; Dom; Sox21; Sox-10	Synonyms	GenebankID or UniProtKB
		XP_005514744 (https://www.ncbi.nlm.nih.gov/nuccore/XP_005514744)
10090.ENSMUSP00000039466 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000039466)	String	
-	Sequence Similarities	
	GO - Molecular Function	
GO:0042802 : identical protein binding (https://www.ebi.ac.uk/QuickGO/term/GO:0042802)		
GO:0001228 : DNA-binding transcription activator activity, RNA polymerase II-specific (https://www.ebi.ac.uk/QuickGO/term/GO:0001228)		
GO:0003700 : DNA-binding transcription factor activity		

(<https://www.ebi.ac.uk/QuickGO/term/GO:0003700>)
GO:0008134 : transcription factor binding
(<https://www.ebi.ac.uk/QuickGO/term/GO:0008134>)
GO:0044212 : transcription regulatory region DNA binding
(<https://www.ebi.ac.uk/QuickGO/term/GO:0044212>)
GO:0003677 : DNA binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0003677>)
GO:0003682 : chromatin binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0003682>)
GO:0000981 : DNA-binding transcription factor activity, RNA polymerase II-specific
(<https://www.ebi.ac.uk/QuickGO/term/GO:0000981>)
GO:0000978 : RNA polymerase II proximal promoter sequence-specific DNA binding
(<https://www.ebi.ac.uk/QuickGO/term/GO:0000978>)
GO:0000980 : RNA polymerase II distal enhancer sequence-specific DNA binding
(<https://www.ebi.ac.uk/QuickGO/term/GO:0000980>)
GO:0003705 : transcription factor activity, RNA polymerase II distal enhancer sequence-specific binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0003705>)
GO:1990841 : promoter-specific chromatin binding
(<https://www.ebi.ac.uk/QuickGO/term/GO:1990841>)

GO - Biological Process

GO:0007417 : central nervous system development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0007417>)
GO:0043066 : negative regulation of apoptotic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0043066>)
GO:0007422 : peripheral nervous system development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0007422>)
GO:0045944 : positive regulation of transcription by RNA polymerase II
(<https://www.ebi.ac.uk/QuickGO/term/GO:0045944>)
GO:0006355 : regulation of transcription, DNA-templated
(<https://www.ebi.ac.uk/QuickGO/term/GO:0006355>)
GO:0030154 : cell differentiation (<https://www.ebi.ac.uk/QuickGO/term/GO:0030154>)
GO:0045892 : negative regulation of transcription, DNA-templated
(<https://www.ebi.ac.uk/QuickGO/term/GO:0045892>)
GO:0001755 : neural crest cell migration
(<https://www.ebi.ac.uk/QuickGO/term/GO:0001755>)
GO:0045893 : positive regulation of transcription, DNA-templated
(<https://www.ebi.ac.uk/QuickGO/term/GO:0045893>)
GO:0090090 : negative regulation of canonical Wnt signaling pathway
(<https://www.ebi.ac.uk/QuickGO/term/GO:0090090>)
GO:0001701 : in utero embryonic development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0001701>)
GO:0010628 : positive regulation of gene expression
(<https://www.ebi.ac.uk/QuickGO/term/GO:0010628>)
GO:0030318 : melanocyte differentiation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0030318>)
GO:0048546 : digestive tract morphogenesis
(<https://www.ebi.ac.uk/QuickGO/term/GO:0048546>)
GO:0002009 : morphogenesis of an epithelium
(<https://www.ebi.ac.uk/QuickGO/term/GO:0002009>)
GO:0048484 : enteric nervous system development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0048484>)
GO:0048469 : cell maturation (<https://www.ebi.ac.uk/QuickGO/term/GO:0048469>)
GO:0032808 : lacrimal gland development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0032808>)
GO:0048589 : developmental growth
(<https://www.ebi.ac.uk/QuickGO/term/GO:0048589>)
GO:0071393 : cellular response to progesterone stimulus
(<https://www.ebi.ac.uk/QuickGO/term/GO:0071393>)
GO:0022010 : central nervous system myelination
(<https://www.ebi.ac.uk/QuickGO/term/GO:0022010>)
GO:0061138 : morphogenesis of a branching epithelium
(<https://www.ebi.ac.uk/QuickGO/term/GO:0061138>)
GO:0010626 : negative regulation of Schwann cell proliferation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0010626>)
GO:0014003 : oligodendrocyte development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0014003>)
GO:0048709 : oligodendrocyte differentiation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0048709>)
GO:0014015 : positive regulation of gliogenesis
(<https://www.ebi.ac.uk/QuickGO/term/GO:0014015>)
GO:0031643 : positive regulation of myelination
(<https://www.ebi.ac.uk/QuickGO/term/GO:0031643>)
GO:0002052 : positive regulation of neuroblast proliferation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0002052>)
GO:0006368 : transcription elongation from RNA polymerase II promoter
(<https://www.ebi.ac.uk/QuickGO/term/GO:0006368>)

GO - Cellular Component

GO:0005737 : cytoplasm (<https://www.ebi.ac.uk/QuickGO/term/GO:0005737>)
GO:0005654 : nucleoplasm (<https://www.ebi.ac.uk/QuickGO/term/GO:0005654>)
GO:0005634 : nucleus (<https://www.ebi.ac.uk/QuickGO/term/GO:0005634>)

GO:0000785 : chromatin (<https://www.ebi.ac.uk/QuickGO/term/GO:0000785>)
GO:0031315 : extrinsic component of mitochondrial outer membrane
(<https://www.ebi.ac.uk/QuickGO/term/GO:0031315>)

No (<https://www.gephebase.org/search-criteria?/and+Presumptive Null=~No~#gephebase-summary-title>)

Cis-regulatory (<https://www.gephebase.org/search-criteria?/and+Molecular Type=~Cis-regulatory~#gephebase-summary-title>)

Deletion (<https://www.gephebase.org/search-criteria?/and+Aberration Type=~Deletion~#gephebase-summary-title>)

1-10 kb

melanocyte conserved enhancer deletion

Linkage Mapping (<https://www.gephebase.org/search-criteria?/and+Experimental Evidence=~Linkage Mapping~#gephebase-summary-title>)

Epistatic and combinatorial effects of pigmentary gene mutations in the domestic pigeon. (2014) (<https://pubmed.ncbi.nlm.nih.gov/24508169>)

Domyan ET; Guernsey MW; Kronenberg Z; Krishnan S; Boissy RE; Vickrey AI; Rodgers C; Cassidy P; Leachman SA; Fondon JW; Yandell M; Shapiro MD

Understanding the molecular basis of phenotypic diversity is a critical challenge in biology, yet we know little about the mechanistic effects of different mutations and epistatic relationships among loci that contribute to complex traits. Pigmentation genetics offers a powerful model for identifying mutations underlying diversity and for determining how additional complexity emerges from interactions among loci. Centuries of artificial selection in domestic rock pigeons (*Columba livia*) have cultivated tremendous variation in plumage pigmentation through the combined effects of dozens of loci. The dominance and epistatic hierarchies of key loci governing this diversity are known through classical genetic studies, but their molecular identities and the mechanisms of their genetic interactions remain unknown. Here we identify protein-coding and cis-regulatory mutations in *Tyrp1*, *Sox10*, and *Slc45a2* that underlie classical color phenotypes of pigeons and present a mechanistic explanation of their dominance and epistatic relationships. We also find unanticipated allelic heterogeneity at *Tyrp1* and *Sox10*, indicating that color variants evolved repeatedly through mutations in the same genes. These results demonstrate how a spectrum of coding and regulatory mutations in a small number of genes can interact to generate substantial phenotypic diversity in a classic Darwinian model of evolution.

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6 (MC1R, Mlana, ndp (norrin), slc2a11b, SLC45A2=MATP, tyrosinase-related protein 1 (TYRP1)) (<https://www.gephebase.org/search-criteria?/or+Taxon ID=~8932~/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title>)

1 (<https://www.gephebase.org/search-criteria?/or+Gene Gephebase=~SOX10~/and+Taxon ID=~8932~/or+Gene Gephebase=~SOX10~/and+Taxon ID=~8932~#gephebase-summary-title>)

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Presumptive Null

Molecular Type

Aberration Type

Deletion Size

Molecular Details of the Mutation

Experimental Evidence

Main Reference

Authors

Abstract

Additional References

RELATED GEPHE

Related Genes

Related Haplotypes

EXTERNAL LINKS

COMMENTS

@Epistasis - Enhancer deletion - Two alleles @AllelicSeries